# EPX gene

eosinophil peroxidase

## Normal Function

The *EPX* gene provides instructions for making a protein called eosinophil peroxidase. This protein is found within certain white blood cells called eosinophils. During a normal immune response, eosinophils are activated (turned on), and they travel to the area of injury or inflammation. The cells then release proteins and other compounds that have a toxic effect on severely damaged cells or invading organisms. One of these proteins is called eosinophil peroxidase. This protein helps form molecules that are highly toxic to bacteria and parasites. These toxic molecules also play a role in regulating inflammation by fighting microbial invaders.

The eosinophil peroxidase protein is produced as a long strand that is cut (cleaved) into two smaller pieces. The shorter piece is known as the light chain and the longer piece is known as the heavy chain. These two pieces are attached to each other to form functional eosinophil peroxidase.

## Health Conditions Related to Genetic Changes

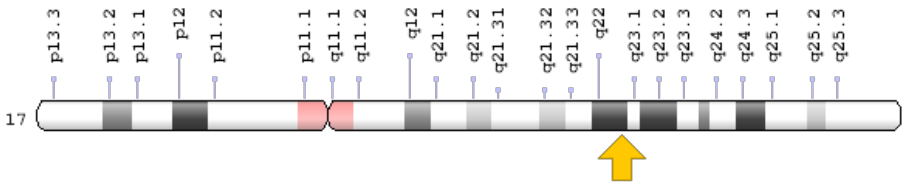
### eosinophil peroxidase deficiency

At least three mutations have been found to cause eosinophil peroxidase deficiency. This condition affects eosinophils but causes no health problems in affected individuals. These mutations reduce or prevent eosinophil peroxidase production or result in a protein that is unstable and nonfunctional. As a result, eosinophils have severely reduced amounts of eosinophil peroxidase or none at all. The cells lacking eosinophil peroxidase are smaller and may have structural changes, but the other proteins within affected eosinophils are normal and the loss of eosinophil peroxidase does not appear to impair the function of these cells.

## Chromosomal Location

Cytogenetic Location: 17q22, which is the long (q) arm of chromosome 17 at position 22

Molecular Location: base pairs 58,192,724 to 58,205,174 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- eosinophil peroxidase preproprotein
- EPO
- EPP
- EPX-PEN

## Additional Information & Resources

Educational Resources

- Immunobiology: The Immune System in Health and Disease (fifth edition, 2001): Eosinophils are Normally Under Tight Control to Prevent Inappropriate Toxic Responses
  https://www.ncbi.nlm.nih.gov/books/NBK27112/#A1739

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EPX%5BTI%5D%29+OR+%28eosinophil+peroxidase%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- EOSINOPHIL PEROXIDASE
  http://omim.org/entry/131399

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_EPX.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EPX%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3423
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8288
- UniProt
  http://www.uniprot.org/uniprot/P11678

## Sources for This Summary


- Acharya KR, Ackerman SJ. Eosinophil granule proteins: form and function. J Biol Chem. 2014 Jun 20;289(25):17406-15. doi: 10.1074/jbc.R113.546218. Epub 2014 May 6. Review.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/24802755
  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4067173/
- OMIM: EOSINOPHIL PEROXIDASE
  http://omim.org/entry/131399
- Romano M, Baralle FE, Patriarca P. Expression and characterization of recombinant human eosinophil peroxidase. Impact of the R286H substitution on the biosynthesis and activity of the enzyme. Eur J Biochem. 2000 Jun;267(12):3704-11.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/10848988
- Yamaguchi Y, Zhang DE, Sun Z, Albee EA, Nagata S, Tenen DG, Ackerman SJ. Functional characterization of the promoter for the gene encoding human eosinophil peroxidase. J Biol Chem. 1994 Jul 29;269(30):19410-9.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/8034708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